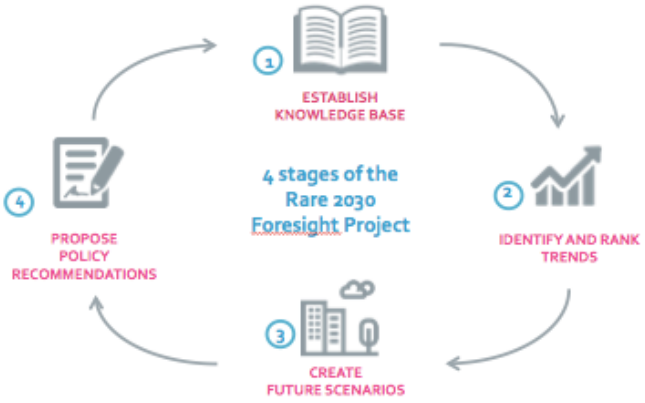
Summary and Preliminary Outcomes of the Rare 2030 EU level back-casting Workshops and Plenary Conference on the Future Vision of European Reference Networks

Contents

Introduction

Rare 2030 is a foresight study requested by the European Parliament and supported by the European Commission to gather input and propose policy recommendations that will lead us to improved policy and a better future for people living with a rare disease in Europe in the next 10 years and beyond.

As a fourth step in the foresight process a series of consultations and workshops have been organised with the 200+ stakeholders involved in the project to agree on the policies needed to be put in place today to reach the preferred Rare 2030 scenarios of tomorrow.



The Rare 2030 Foresight Study and the resulting recommendations cast a particular focus on the future for European Reference Networks (ERNs), given their central importance to the rare disease community. Rare 2030 partners organised **4 parallel workshops,** divided into four 2.5-hour breakout sessions staggered across several weeks in **September and October 2020**. Each of the four working sessions focused on an area of major strategic interest to ERNs:

21 SEPTEMBER 2020 Governance and Strategic positioning of ERNs.
Should the ERNs have a legal status, and if so, what is the best route/best way to achieve this? How can we secure financial sustainability of ERNs? Future composition of ERNs (in terms of

model and scale): can we reach the right balance between Centres directly and indirectly involved in the ERNs?

28 SEPTEMBER 2020 Integrating ERNs to national systems and frameworks
What is the best way to integrate ERNs into national health systems? How should ERNs complement the wider national landscape of Centres of Expertise for RD? What role do you see ERNs playing in bridging health and social care? Should ERNs drive future policies at national level, and if so, how?

29 SEPTEMBER 2020 Role of ERNs in virtual care delivery and cross-border healthcare
How can the CPMS transform virtual care for specialised conditions, and how can the ERNs more widely accelerate positive telemedicine trends towards balanced physical-virtual clinics of the future? What does success look like for you? How can we receive legal/regulatory/financial recognition of time and expertise spent on cross-site CPMS case discussions? Could/should decisions of ERN panels bear more weight?

12 OCTOBER 2020 ERNs, research, and the data ecosystem of the future
What should be the ERNs' 'data strategy' in 2030? How can ERNs contribute to diagnostic equality across Europe? What is the best, most realistic role for them to play? How do we want ERNs to be positioned, research-wise? How do we review the policies (and address the problems) around data-sharing and health & research?

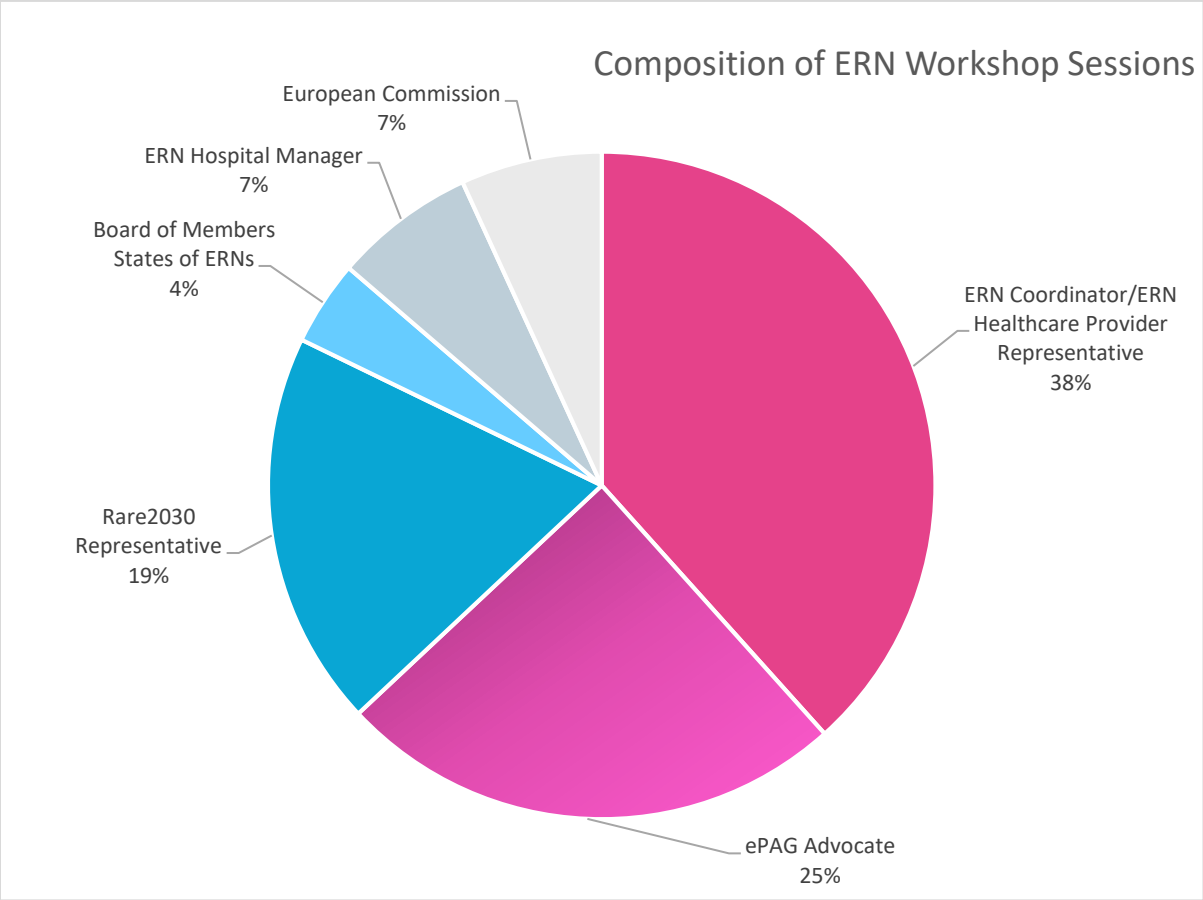
Summary of the Working Sessions

The 4 breakout sessions together involved 73 participants, including ERN representatives, ERN Hospital Managers, ePAG advocates, EURORDIS experts, Board of Members States of ERNs representatives, key members of the European Commission, and partners of the Rare2030 project.

The breakdown of stakeholders was as follows:

ERN Coordinator/ERN Healthcare Provider Representative	28
ePAG Advocate	18
Rare2030 Representative	14
Board of Members States of ERNs	3
ERN Hospital Manager	5

European Commission	5



Each session centred on a number of proposed preliminary recommendations directed towards ERNs. **In total, across the 4 sessions of the back-casting workshop, over 80 preliminary recommendations were discussed.**

Participants spent some of the session time reviewing and ranking these preliminary recommendations individually, and the rest of the time working in groups, to identify the recommendations the participants considered to be of greatest strategic importance to ERNs fulfilling their potential in 2030. The groups also brainstormed on the feasibility of each recommendation, with the reminder to think ambitiously. Wherever possible, the participants proposed steps which would be needed to implement some of these strategic recommendations.

Plenary Event

These working sessions were followed by a **Closing Plenary on 26 October 2020,** to present the main conclusions from all sessions and open up participation to a wider audience. After the final working session, the recommendations deemed most relevant had been extracted - and amended where necessary - and were incorporated to a survey (featuring 33 recommendations in total – see below). This survey asked respondents to consider all 33 and rank them from 1-10 in terms of strategic importance to the future of ERNs. In this way, the Rare 2030 partners would be able to identify more objectively which recommendations should be prioritised in the policy-making activities of the coming months and years. The survey was disseminated to all workshop participants, and the preliminary results were shared during the Plenary event, to enable experts to provide their reactions and responses to these recommendations and how to implement them.

Following the Plenary, the recommendations survey remained open to participations, but was also shared more widely with the broader rare disease community, in particular through ePAG communication channels, through individual ERN communications to their respective centres, and to the partners of the European Joint Programme for Rare Disease Research.

Full list of Recommendations included in the Survey

The recommendations included to this survey can be viewed below (grouped by strategic area, in line with the four working sessions)

SESSION 1 – GOVERNANCE AND STRATEGIC POSITIONING OF ERNS

1. ERNs need a long-term funding framework which should consider ALL possible sources of funding: such a framework needs to be defined urgently and should include a definition of all central functionalities and policies to support ERNs' financial management and governance

2. The realistic costs of network coordination, relative to the activities of the ERNs, should be established, and coordination funding provided on these grounds – the amount of funding available for coordination-related activities should differ between ERNs, based on scale and coverage

3. A common EU agency should be created/adapted to enable ERNs to operate more flexibly and effectively, and receive funding from a range of sources (including 'external' sources such as industry and private donors, with an appropriate governance for public-private partnerships)

4. ERNs should ideally each be legal entities – in the interim, they should be nested within an organisation such as a foundation, to provide a mechanism for ERNs to easily receive funds.

5. Countries should be encouraged to revisit and update their national designation of CEs for rare diseases and strengthen the organisation of national rare disease and specialised care networks - this should then translate to a more strategic engagement of national CEs with ERNs, via a limited number of full member HCPs.

6. ERNs should develop clear and transparent rules for patient engagement, adequately supporting the involvement of patient organisations and their representatives in the different ERN activities and fairly compensate patient representatives
7. The EU (EC) should provide coordination funding to coordinating HCPs but MS/EEA countries should provide a small amount of money to each national HCP/affiliated member within their national territory (providing they meet performance and impact indicators) to support their engagement in ERN activities

8. A special category of association or collaboration or affiliation should be created to allow formal collaboration and recognition of centres from countries outside of the EU MS/EEA; clear rules on shared activities (what is and is not permitted) should be created.

SESSION 2 – INTEGRATING ERNS TO NATIONAL SYSTEMS AND FRAMEWORK

9. The European Commission should support Member States/EEA countries to implement the actions outlined in the ERN BoMs Statement on Integration of ERNs into national health systems, specifically by funding national multi-stakeholder workshops, with patient organisations, clinical leads and national authorities, to facilitate discussions and actions on integration into each of the national health systems.

10. The concept of a centre of expertise for rare diseases should be revisited at country level: countries should ensure they designate all such centres in a comprehensive and transparent way, and make the result of such a mapping and designation publicly available, demonstrating how ERN HCPs and 'affiliated' centres fit within wider national networks (where applicable). The *EUCERD Quality Criteria for Centres of Expertise for Rare Diseases* remain a robust resource here

11. To facilitate the referral of patients for ERN care, all Member States/EEA countries should endorse one centre as a 'National Coordination Hub', to function as a central referral management centre and gateway to accessing the specialist advice of the ERNs collectively: this centre should work in partnership with the national patient community, and build relationships with national professional societies and the research leads

12. National competent authorities should define national referral pathways for rare disease (or suspected rare disease) patients or those requiring a concentration of expertise, addressing transition from paediatric to adult care and containing clear guidance on how and when to seek referral to an ERN; ERNs should then compile and publish these pathways, explaining the process in each country and producing -with its patient advocates- patient-friendly information and advice on accessing specialist advice under an ERN.
13. ERNs should be supported to review and expand their disease-specific membership criteria, in collaboration with patients and professional associations, with an emphasis on the necessary multidisciplinary expertise: in this way, EU countries (perhaps even the global RD community) could make use of robust criteria by which to define expertise in given disease areas

14. ERNs and the BoMS should embrace a strategic mission of promoting more integrated care, encompassing integration of different medical specialities, but also of paramedical and social actors, in line with the *EUCERD Recommendations on Rare Disease European Reference Networks* and the *Commission Expert Group Recommendations to support the incorporation of rare diseases to social policies and services*

15. By 2025 a cross-ERN working group on integrated and holistic care should have been established in partnership with European Resources Centres for Rare Diseases, as a gateway to build joint guidance on collaborative approaches for the provision of integrated and holistic care to people living with a rare disease: dedicated funding should be made available for broad stakeholder meetings and activities to advance this goal

16. ERNs should gather and create, in collaboration with patient organisations, resources which could support rare disease patients in receiving more integrated and more personalised care in their local environment: such resources should translate to heterogenous care and social settings, by focusing on clarifying and explaining the (often poorly-understood) needs of patients with complex conditions, and adaptations/approaches which could help

17. Renewing or updating national plans and strategies for rare diseases should remain a key priority for all countries - all such documents should stipulate the strategy to engage bidirectionally with ERNs, and support in this task should be provided by a group/body with a remit to encompass all RD topics, beyond ERNs alone

18. Each Member State/EEA should define a mechanism to disseminate and utilise the knowledge and evidence generated by the ERNs, to impact across the wider health and social systems: in particular, clinical practice guidelines/clinical decision support tools generated or endorsed by an ERN should be fully applied in all MS/EEA countries, and national committees and structures dedicated to rare disease or specialised healthcare should include some level of national ERN HCP representation

SESSION 3 – ROLE OF ERNS IN VIRTUAL CARE DELIVERY AND CROSS-BORDER HEALTHCARE

19. The CPMS should be fully compatible with any referring HCP systems, enabling automatic and two-way cross-talk with Electronic Health Records to populate and update records post case referral: data should be fully searchable, and cases accompanied by an appropriate PPRL (privacy preserving record linkage) solution to facilitate integration of ERNs

20. A dedicated study/project should be funded, to support countries in developing their EHRs and virtual care delivery services to best address the specificities of rare diseases and highly specialised healthcare, and promote interoperability - this could aim at wider national deployment of the CPMS or a system compatible with it, as the basis for virtual care provision for complex rare disease cases nationally

21. ERNs should develop strategies to minimise disparity in access to high quality healthcare between European regions - disease-related metrics should be agreed and monitored

22. An efficient project/tender should be funded to establish a pricing model to reimburse expert time spent on CPMS case review and propose alternatives for payment (e.g. a quid pro quo system, a straightforward

billing of another MS/EEA country - perhaps with a differential GDP-based pricing scheme- a reduction in the workload of ERN HCP clinicians in lieu of payment for CPMS reviews, etc)

23. The social security regulation/ cross-border healthcare directive should be amended to allow for payment of time spent on cross-border virtual consultations performed through the CPMS, following a systematic national referral process

24. Countries should automatically authorise requests for treatments or therapies if deemed beneficial by an ERN panel through CPMS: the ERNs' expertise should hold more weight than national expert bodies who make such decisions as present

SESSION 4 – ERNS, RESEARCH, AND THE DATA ECOSYSTEM OF THE FUTURE

25. The future ERN health data strategy must be anchored to the wider European health data and IT ecosystem, driven by a concerted policy action of all the relevant DGs and aligned with national health data strategies from the majority of MS/EEA countries; in this context, ERNs should help to shape the future Health Code of Conduct for secondary use of data (addressing the need to make GDPR research-friendly)

26. The future ERN data strategy must be targeted towards all rare disease patients in Europe, and not only those attending ERN HCPs: opportunities must be created for patients to foster robust data partnerships, determine governance, and contribute/extract data to or from appropriate registries, care records and other relevant data sources.

27. By 2023, ERNs should develop a comprehensive data strategy and implementation plan envisaging the necessary activities across 6 action lines: architecture - cloud computing services and IT support for registries and other databases; data collection protocols; data curation services; data management tools (services and tools to search, access and share data, tools to manage own data); data analytics tools and services; and a data governance framework.

28. Data from hospital EHR (electronic health record) systems should be interoperable with the CPMS, and with ERNs' new epidemiological registries, allowing minimal data entry and maximum automation (with accompanying quality assurance)

29. Disease-specific registries (where positively evaluated by ERNs based on rigorous criteria OR created anew by ERNs in future) should be interoperable with the new ERN registries and any robust national RD registries: these should all be connected (sustainably) to ERDRI* to provide a fully functioning registration ecosystem

30. ERNs should sit at the centre of all future efforts to refine and evolve all ontologies and standards for data collection and utilisation into a common data model, including efforts to facilitate extraction and mining from real-world data

31. ERNs should receive earmarked -and adequate- funding through European programmes to deliver clinical trials (involving centres inside or outside of the Networks, as required) and to research neglected topics including rehabilitative, holistic and social research

32. ERNs should be specifically and adequately funded to develop and conduct natural history (and where possible accompanying biomarker)studies, a minimum of 5 every 2 years, to build the knowledge base and capacity for clinical research in neglected diseases/areas lacking research

33. The Coordination and Support Action funded by the H2020 programme to support the creation of Clinical Research Networks (covering 4 domains: clinical research; data management; engagement and dissemination; and administrative support) should be supplemented by additional funding to deploy core services to become fully operational by 2025

Preliminary Results of the Survey

In total, 204 complete responses to the recommendations survey were received. **The ten recommendations deemed to be of greatest strategic importance for the future of ERNs are as follows (in descending order)**

Recommendation	**Score in Survey**
ERNs need a long-term funding framework which should consider ALL possible sources of funding: such a framework needs to be defined urgently and should include a definition of all central functionalities and policies to support ERNs' financial management and governance	**1791**
The concept of a centre of expertise for rare diseases should be revisited at country level: countries should ensure they designate all such centres in a comprehensive and transparent way, and make the result of such a mapping and designation publicly available, demonstrating how ERN HCPs and 'affiliated' centres fit within wider national networks (where applicable). The *EUCERD Quality Criteria for Centres of Expertise for Rare Diseases* remain a robust resource here	**1604**
Renewing or updating national plans and strategies for rare diseases should remain a key priority for all countries - all such documents should stipulate the strategy to engage bidirectionally with ERNs, and support in this task should be provided by a group/body with a remit to encompass all RD topics, beyond ERNs alone	**1592**
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connected (sustainably) to ERDRI* to provide a fully functioning registration ecosystem	
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Each Member State/EEA should define a mechanism to disseminate and utilise the knowledge and evidence generated by the ERNs, to impact across the wider health and social systems: in particular, clinical practice guidelines/clinical decision support tools generated or endorsed by an ERN should be fully applied in all MS/EEA countries, and national committees and structures dedicated to rare disease or specialised healthcare should include some level of national ERN HCP representation	**1584**
The European Commission should support Member States/EEA countries to implement the actions outlined in the ERN BoMs Statement on Integration of ERNs into national health systems, specifically by funding national multi-stakeholder workshops, with patient organisations, clinical leads and national authorities, to facilitate discussions and actions on integration into each of the national health systems	**1584**
ERNs should receive earmarked -and adequate- funding through European programmes to deliver clinical trials (involving centres inside or outside of the Networks, as required) and to research neglected topics including rehabilitative, holistic and social research	**1583**
The EU (EC) should provide coordination funding to coordinating HCPs but MS/EEA countries should provide a small amount of money to each national HCP/affiliated member within their national territory (providing they meet performance and impact indicators) to support their engagement in ERN activities	**1575**
National competent authorities should define national referral pathways for rare disease (or suspected rare disease) patients or those requiring a concentration of expertise, addressing transition from paediatric to adult care and containing clear guidance on how and when to seek referral to an ERN; ERNs should then compile and publish these pathways, explaining the process in each country and producing -with its patient advocates- patient-friendly information and advice on accessing specialist advice under an ERN	**1558**

Next Steps

The final results will be available by January 2021, via a separate 'ERN Recommendations Survey' document. Furthermore, key findings will be included in the broader set of recommendations resulting from the Rare 2030 Foresight Study, which will be presented to European and national policy makers and key opinion leaders during the **Rare 2030 Final Online Conference 23 February 2021**, with recommendations on the most critical areas requiring robust policies.

Please visit www.rare2030.eu for more information on the project and how to join the final event.